INTRODUCTION

Students in genetics classes become familiar with interpreting pedigrees for modes of inheritance of single-gene traits. Non-majors' genetics courses often have students produce pedigrees of their own families. This exercise provides students with an extensive database, including over 50 families, from which they are asked to produce pedigrees as evidence in support of or refuting suggested models of inheritance for simple traits. Students are also asked to propose modes of inheritance for complex human traits based on pedigrees that they produce from the database. The people in the database are real and include their own families. This makes the exercise more real and personal for the students.

The exercise is used to teach about inheritance, introduce students to the problems encountered when collecting categorical phenotypic data, and provide practice at evaluating data in order to identify appropriate and informative data from a large dataset that may be used to answer a particular question. Using a database of this size, though not large for some of us, helps to give our students the experience suggested by Vision and Change in Undergraduate Biology (1).

This teaching tool was developed for use in an introductory, sophomore-level genetics class. Due to the extensive nature of the database, it is used as an exercise for a three-hour lab. It can be modified for use in an Introductory Biology course or a non-majors' genetics class.

PROCEDURE

For the past four years, students in the biology majors' genetics class at Alfred University have been required to collect phenotypic data of their families. The information collected includes: gender, relationship to others, age, eye color, handedness, hair color, height, heart disease (yes or no), and hypertension (yes or no). Students are given a datasheet in two forms: 1) as a handout (Supplement 1 Genetic Information Data Sheet), and 2) as an Excel spreadsheet (Supplement 2 Family Database; Data Sheet for Students tab). Four lines of data are provided in the datasheet as an example for how to describe family relationships. Students submit their data in Excel format so that it can be easily added to the larger database (Supplement 2 Family Database; Database tab). In the compiled database, the data are labeled with data collection year, a family code, and the initials of each individual in the database. No names are used. The current database includes 672 individuals from 56 families, ranging in age from less than one year to 93 years old. The traits could be expanded to include presence (dominant) or absence (recessive) of freckles or any other simple or complex human traits that are easy to phenotype.

The database is introduced to students in a three-hour lab session, during which they are guided through the process of sorting, selecting, and organizing data for analysis. They will find that some data is missing and some is not in the expected format, but dealing with that is part of the learning process. Students then have access to the compiled database on-line and are able to sort and select the information that they need to answer the following questions. In their reports, they are required to provide pedigrees to support their answers and to describe the criteria they used for the selection of the specific families that they have included in their reports.

Questions for upper-level students

1. Does our data support the current models of inheritance for handedness and eye color? Explain.
2. Hypothesize about the mode of inheritance for hair color, heart disease, and hypertension. Provide the data in support of and/or refuting your hypothesis. What additional information would you like to further test your hypothesis?
3. Do you detect linkage between any of the traits in the database? For example, do blue eyes and blonde hair occur together more often than would be expected due to independent assortment? (Note: This is a more advanced question.)

Assignments for introductory biology or non-majors' genetics classes

1. Identify two families in the database that show variation for one trait and produce pedigrees for those families.
2. Assign genotypes for each individual based on the current model for inheritance of that trait.
EMMONS: SIMPLE AND COMPLEX TRAITS USING PEDIGREES

CONCLUSION

Students in the most recent genetics class were asked: 1) What one or two things did you learn from this lab exercise?, and 2) What would you change about the exercise? Nearly every student reported a better understanding of inheritance patterns from comparison of family pedigrees. A number of students indicated that they learned that inheritance of simple traits does not always follow the “black and white rules” as presented in class, and the analysis often “produced more questions than answers.” The majority of students found using Excel to manipulate the data to be challenging. Some acknowledged that now they can use the program “better” and “for many useful things.” Nearly every student suggested allowing more time for the analysis (they were given two weeks after the data was introduced in lab). Every student was able to submit an acceptable report, with pedigrees in support of their answers to the questions in the time allowed. The time allowed should be determined based on student abilities. The students had some difficulty determining relationships among individuals in some families and did not like that some data were provided in non-standard formats or were missing. That point was used to emphasize the need for accurate data collection.

Teachers’ notes

Handedness: The genetics of handedness has been partially described. The allele for right-handedness is considered to be dominant to the allele for left-handedness and there is no current genetic explanation for ambidextrousness (8). Database phenotypes include right, left, and ambidextrous. There are only 10 families with at least one ambidextrous member and only one family with two ambidextrous members.

Eye Color: Two genes have been identified that explain the occurrence of brown, green, and blue eyes. The bey2 locus on chromosome 15 (4) has two known alleles for which brown (B) is dominant to blue (b). The gey locus on chromosome 19 (3) has two known alleles for which green (G) is dominant to blue (g). Brown alleles at bey2 are epistatic to the green alleles at gey, producing brown eyes. Therefore only one brown allele at bey2 is necessary to produce brown eyes (B_:_ __). Four blue alleles are required to produce blue eyes, two at each locus (bb;gg). Green eyes are produced if there are two blue alleles for bey2 and at least one green allele for gey (bb;G_). There is no current genetic explanation for gray, purple, or hazel eyes. Eye color phenotypes for the database are limited to brown (includes hazel), blue (gray, purple, etc.), and green. There are 35 families in the database with at least one member with green eyes.

Hair Color: A complete genetic model explaining the inheritance of hair color is not available (7). It has been proposed that red hair is due to a number of recessive alleles of the MC1R locus (9). Hair color phenotypes for this database are limited to black, brown, blonde, and red, to simplify the pedigrees. This is the most difficult trait for getting the students to use only the allowed phenotypes. They also may have difficulty distinguishing between black and dark brown. You may want to set criteria using color samples from hair dye boxes. The database contains 21 families with at least one red-haired member.

Disease: Heart disease and hypertension are complex traits (2, 6). Susceptibility to these diseases has a genetic basis but is also highly influenced by environmental factors. When choosing families and interpreting the pedigrees for heart disease and hypertension it is important for the students to consider the ages of family members.

More advanced students can analyze the height data for families (complex trait) using regression analysis or the trend line function of Excel to estimate the heritability of height (degree to which the trait is due to genes rather than environment). Students can learn to produce XY scatter grams of offspring height as a function of the mean height of the parents. The slope of the trend line is the heritability of the trait (5).

SUPPLEMENTARY MATERIALS

Genetic Information Data Sheet
Family Database

REFERENCES